

## Supplementary Tables

**Supplementary Table S1. Primer Sequences**

Primer Name	Sequence (5'-3')	Figure
CRB1-Exon 3-forward	GCTGTGTGTGGATGGAGAAA	Fig. 4A
CRB1-Exon 7-reverse	CAGTCTCTGCTTGTCAAGTAG	Fig. 4A
CRB1-Exon 7-forward	CAGGATGACTCCACTGGTTATG	Fig. 4B
CRB1-Exon 8-reverse	TGAATCCACACCACTGAACC	Fig. 4B
CRB1-Exon 6-forward	GTGAAGCTGGAGCTGCTAAGTG	Supp. Fig. S3
CRB1-Exon 6-7-reverse	GCCACATACTCTCAGACAGTTG	Supp. Fig. S3
GAPDH-forward	AGAAGGCTGGGGCTCATTTG	Fig. 3M
GAPDH-reverse	AGGGGCCATCCACAGTCTTC	Fig. 3M
PAX6-forward	CTGAGGAATCAGAGAACAGAGC	Fig. 3M
PAX6-reverse	ATGGAGCCAGATGTGAAGGAGG	Fig. 3M
RCVRN-forward	CCAGAGCATCTACGCCAAGTT	Fig. 3M
RCVRN-reverse	CCGTCGAGGTTGGAATCGAAG	Fig. 3M
CRB1-Exon 2-forward	TGGGACAATCTGTGAAACTACC	Fig. 3M
CRB1-Exon 2-reverse	GGCAGATGCAGACAGGATAA	Fig. 3M

**Supplementary Table S2. Pathogenicity classification for *CRB1* variants**

Variant	Predicted protein description	ACMGG/AMP Classification	Evidence
<i>CRB1</i> c.1892A>G	p.(Tyr631Cys)	Pathogenic (iiia)	PS3* PM2 PM3 PM(>PP1) <sup>#</sup>
<i>CRB1</i> c.2548G>A	p.(Gly850Ser)	Pathogenic (iiia)	PS3 PM2 PM3 PM5 PP3
<i>CRB1</i> c.1892A>G (revised)	p.(Gly391_Arg709del)	Pathogenic (ia)	PVS1 PS3* PM2 PM3 PM(>PP1) <sup>#</sup>

\*PS3 – based on (Quinn et al., 2019)

<sup>#</sup>PM(>PP1) identifies an upgrade from supporting (PP1) to moderate evidence, based on the presence or absence of this variant in one unaffected and two affected siblings from 1 pedigree (N=1/16) (Jarvik & Browning, 2016).

Jarvik, G. P., & Browning, B. L. (2016). Consideration of Cosegregation in the Pathogenicity Classification of Genomic Variants. *Am J Hum Genet*, 98(6), 1077-1081.

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Quinn, P. M., Buck, T. M., Mulder, A. A., Ohonin, C., Alves, C. H., Vos, R. M., . . . Wijnholds, J. (2019). Human iPSC-Derived Retinas Recapitulate the Fetal CRB1 CRB2 Complex Formation and Demonstrate that Photoreceptors and Muller Glia Are Targets of AAV5. *Stem Cell Reports*, 12(5), 906-919. doi:10.1016/j.stemcr.2019.03.002